E-Study material
For 3rd Semester Botany Honours (CBCS)
Course Code: BC307T
Core Course VII: Genetics
Unit 1: Mendelian genetics and its extension
Topic: Lethal Gene, Epistasis and Pleiotropy

Debajit Saikia
Assistant Professor
Department of Botany
CNB College, Bokakhat

Intergenic or nonallelic interaction

In the intergenic or nonallelic interaction, two or more independent genes belonging to same or different chromosomes interact to form a different expression, e.g., complementary genes, supplementary genes, duplicate genes, epistasis, lethal genes.

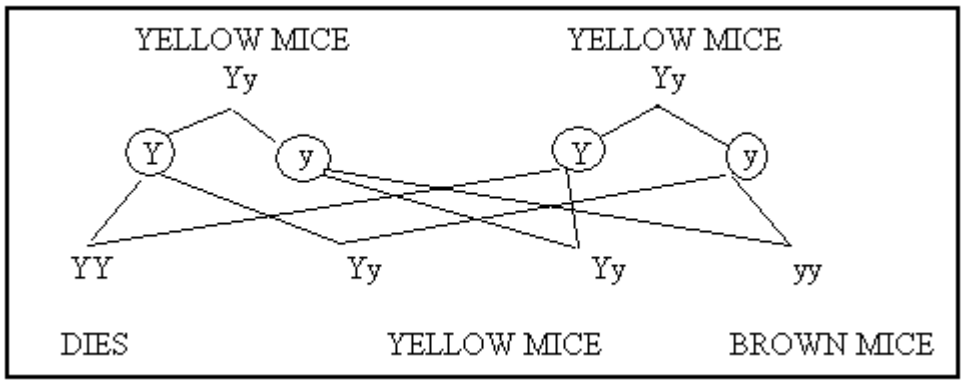
Lethal genes

A lethal gene can be defined as a gene that control some vital functions of the organism and cause death of the organisms in pure recessive or pure dominant form. Lethal genes were first discovered by Cuenot. With the death of homozygous lethal the monohybrid ratio comes to 2 : 1. It may be three types

A. Dominant lethality For eg. Yellow fur in mice.
B. Recessive lethality. For eg sickle cell anaemia.

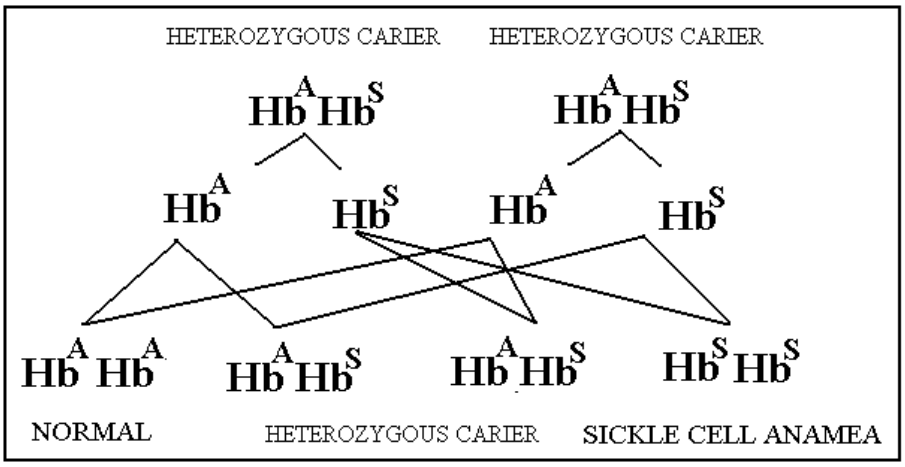
EXAMPLE (1):

When the Yellow mice (Yy) were crossed with Yellow mice (Yy), always Yellow and Brown mice were obtain in the ratio of 2:1. The homozygous dominant mice (YY) dies due to lethality of dominant gene



EXAMPLE (2):

The disease sickle cell anaemia is caused by a recessive gene (Hb^S) which is lethal in homozygous condition. The homozygous for this gene (Hb^SHb^S) generally die of fatal anemea. The heterozygous or cariers for Hb^S i.e. Hb^AHb^S show mild haemolytic anaemea as their R.B.C become sickle shaped and cannot carry large number of Haemoglobin. As a result reduce in oxygen deficiency. C.



<u>Epistasis</u>

Epistasis is the interection between non-allelic genes (present at separate loci) in which one gene maskes, inhibits or supresses the expression of other gene. The gene that suppresses the other gene is known as inhibiting or epistatic factor and the one which is prevented from exhibiting itself is known as *hypostatic*.

OR

Epistasis can be defined as the phenomenon of gene interaction whereby one gene interferes with the phenotypic expression of another non allelic gene or genes. The gene or locus which suppresses or masks the action of a gene at another locus is called epistatic gene. The gene or locus whose expression is suppressed by an epistatic gene is called hypostatic gene.
Epistasis is of three types
Dominant (12:3:1 or 13:3),
recessive (9:3:4) and
dominant-recessive.

Epistatic or Masking Gene Interaction (12:3:1)
In this interaction, the two non-allelic genes affecting the same trait can produce distinct phenotypes alone. But when they are present together, the expression of one gene is masked by the other and a different phenotype is produced when both are present in recessive state. This phenomenon is known as epistasis. It is not a case of inhibition, but the expression of one gene is more intense than the expression of other. The former one is known as epistatic gene and latter one is known as hypostatic gene.

P Black (*BByy*) × Yellow (*bbYY*)

F_1 red purple (*BbYy*)

♀ \ ♂	BY	By	bY	by
BY	*BBYY* Black	*BBYy* Black	*BbYY* Black	*BbYy* Black
By	*BBYy* Black	*BByy* Black	*BbYy* Black	*Bbyy* Black
bY	*BbYY* Black	*BbYy* Black	*bbYY* Yellow	*bbYy* Yellow
by	*BbYy* Black	*Bbyy* Black	*bbYy* Yellow	*bbyy* White

F_2

Phenotypic ratio = 12 Black : 3 Yellow : 1 White

Fig: Epistatic gene interaction showing 12 : 3 : 1 ratio in the development of seed coat colour of barley in F_2

The phenomenon is very analogous to the simple dominance. But they are distinguished by the fact that epistasis is involved among non-allelic genes, whereas Simple dominance is involved among allelic genes. As a result a modified dihybrid ratio 12:3:1 instead of 9:3:3:1 is obtained.

The most epistatic effects are associated with different colour formation in plants and animals.

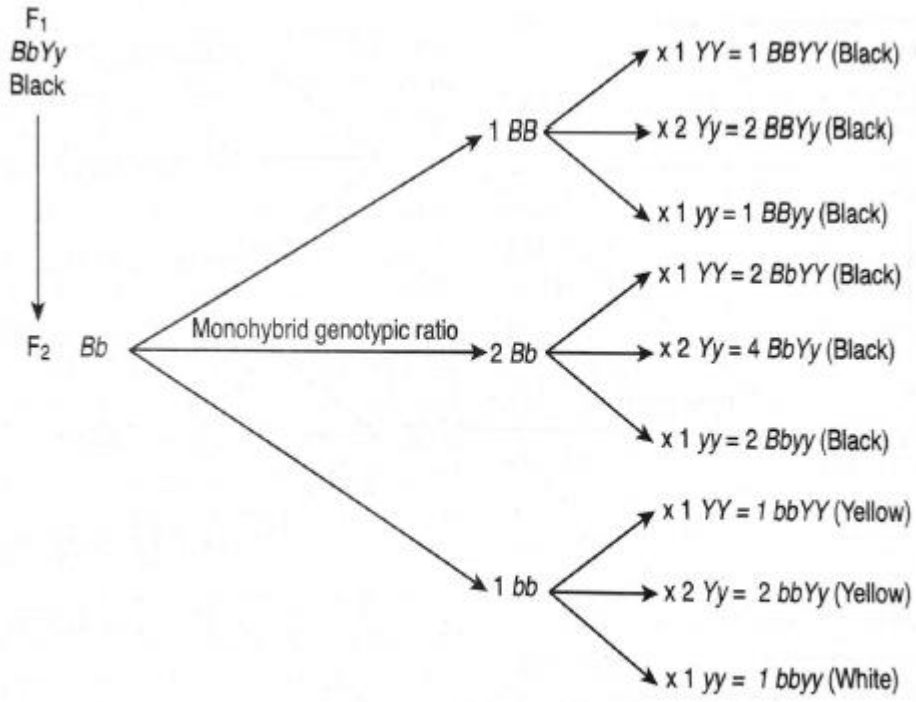


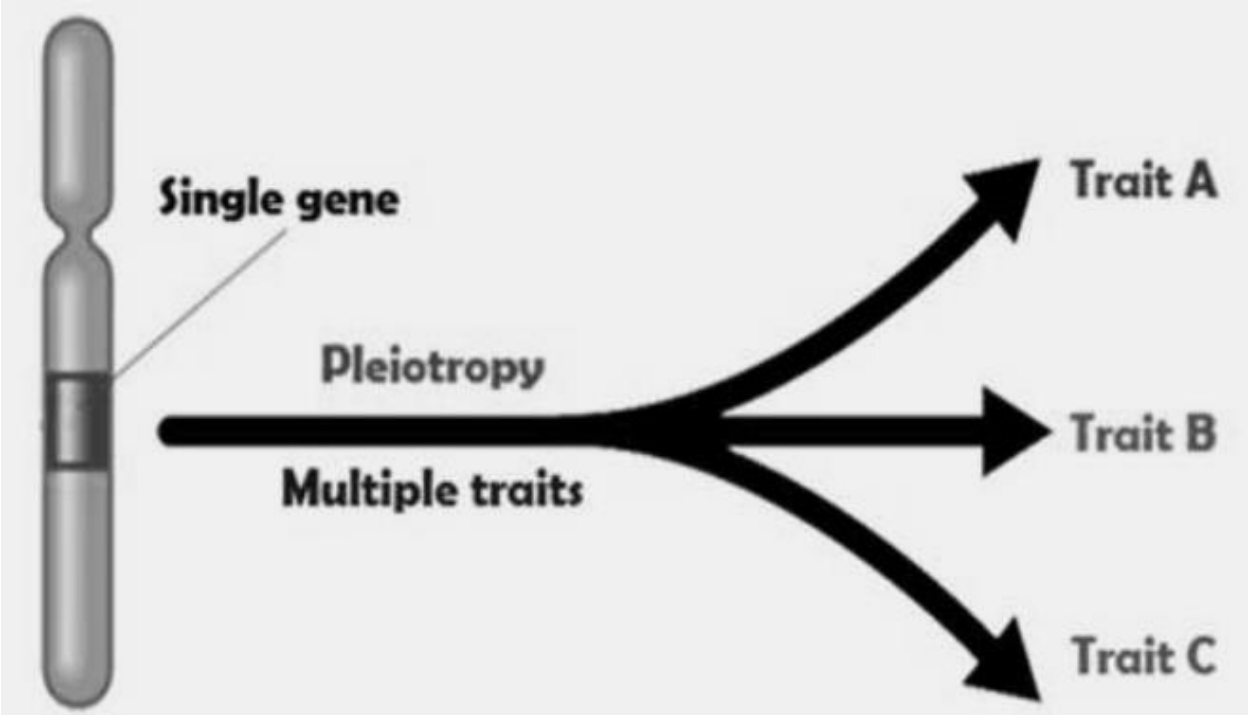
Fig: Epistatic gene interaction showing seed coat colour of barley in 12 : 3 : 1 ratio in F_2 through forked line method

In barley, the seed coat colour is produced by two dominant genes B and Y. Gene B is responsible for black colour. Its homozygous recessive condition produces white seeds. The other gene Y alone is responsible for yellow seed formation, while its homozygous recessive condition also produces white seeds.

When both the dominant alleles are present the phenotype is black seeded plants because the black colour produced by B gene masks the yellow colour produced by Y gene. The dominant gene Y alone produces yellow colouration. When a black seeded strain of the genotype BByy is crossed with yellow seeded strain of the genotype bbYY the F1 (BbYy) produces black seed Coats. In the F2, black, yellow and white seeded plants appear in the 12:3:1 ratio.
A similar interaction is also found in the inheritance of fruit colour in squash and grain colour in sorghum and millet.

Pleiotropy:

They are genes which influence more than one trait. The phenomenon of a single major gene influencing more than one character (multiple expressions) is known as pleiotropic genes. In man gene producing the disease phenylketoneuria also produces a number of phenotypic traits. Besides Sickle cell anemia is another example of pleiotropism.



Pleiotropy in Humans

There are many examples of pleiotropic genes in humans, some of which are associated with disease. For instance, Marfan syndrome is a disorder in humans in which one gene is responsible for a constellation of symptoms, including thinness, joint hypermobility, limb elongation, lens dislocation, and increased susceptibility to heart disease. Similarly, mutations in the gene that codes for transcription factor TBX5 cause the cardiac and limb defects of Holt-Oram syndrome, while mutation of the gene that codes for DNA damage repair protein NBS1 leads to microcephaly, immunodeficiency, and cancer predisposition in Nijmegen breakage syndrome.

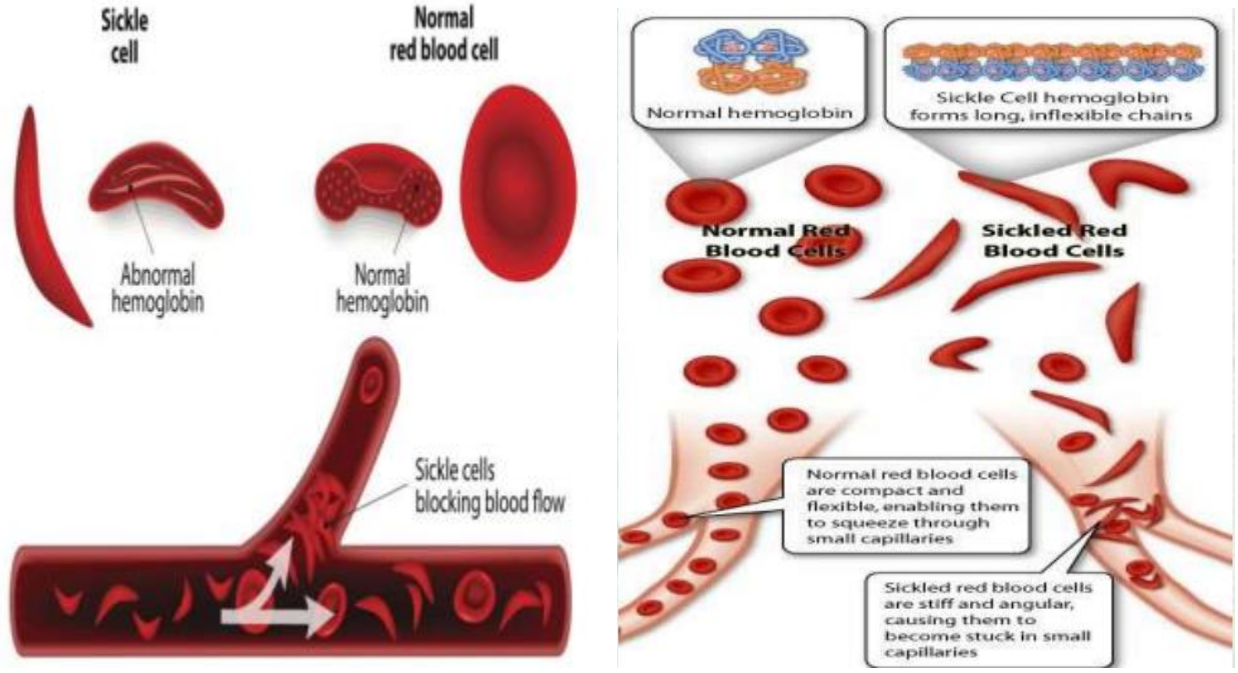
One of the most widely cited examples of pleiotropy in humans is phenylketonuria (PKU). This disorder is caused by a deficiency of the enzyme phenylalanine hydroxylase, which is necessary to convert the essential amino

acid phenylalanine to tyrosine. A defect in the single gene that codes for this enzyme therefore results in the multiple phenotypes associated with PKU, including mental retardation, eczema, and pigment defects that make affected individuals lighter skinned (Paul, 2000).

Sickle cell disease:

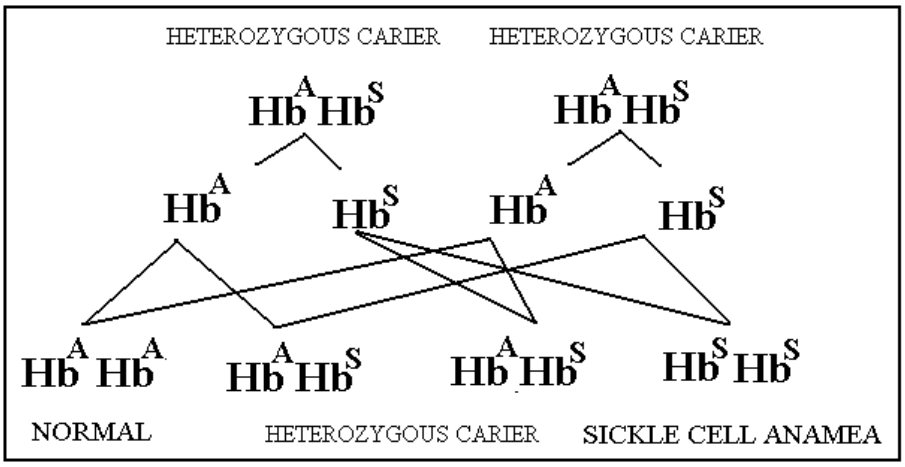
Sickle cell disorder results from the development of abnormally shaped red blood cells. Normal red blood cells have a biconcave, disc-like shape and contain enormous amounts of a protein called hemoglobin.

Sickle cell is a result of a mutation in the beta-globin gene. This mutation results in red blood cells that are sickle-shaped, which causes them to clump together and become stuck in blood vessels, blocking normal blood flow.
The phenotypic effects that single genes may impose in multiple systems often give us insight into the biological function of specific genes.



The disease sickle cell anaemia is caused by a recessive gene (Hb^S) which is lethal in homozygous condition. The homozygous for this gene (Hb^SHb^S) generally die of fatal anemea. The heterozygous or cariers for Hb^S i.e. Hb^AHb^S show mild haemolytic anaemea as their R.B.C become sickle shaped and

cannot carry large number of Haemoglobin. As a result reduce in oxygen deficiency. C.



Pleiotropic genes can also provide us valuable information regarding the evolution of different genes and gene families, as genes are "co-opted" for new purposes beyond what is believed to be their original function (Hodgkin, 1998). Quite simply, pleiotropy reflects the fact that most proteins have multiple roles in distinct cell types; thus, any genetic change that alters gene expression or function can potentially have wide-ranging effects in a variety of tissues

References:

1. A Textbook Of Botany by Ashim Kumar Ghosh, Kashinath Bhattachariya, Gopinath Hait, New Central Book Agency(P) Ltd.,2015, ISBN: 978 81 7381 965 0.
2. Genetics by P.K. Gupta, Rastogi Publications,1997, ISBN: 81-7133-329-X.
3. Cell Biology, Genetics & Molecular Biology By Dipak Kumar Kar, Soma Halder, New Central Book Agency(P) Ltd., 2009, ISBN: 978 81 7381 949 0.